Disease Name — Biotinidase Deficiency

Alternate name(s)	MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET BTD DEFICIENCY
Acronym	BIOT
Disease Classification	Metabolic Disorder
Symptom onset	Prior to 12 months of age
Symptoms	In the untreated state, profound biotinidase deficiency during infancy is usually characterized by neurological and cutaneous findings that include seizures, hypotonia, and rash, often accompanied by hyperventilation, laryngeal stridor, and apnea. Older children may also have alopecia, ataxia, developmental delay, neurosensory hearing loss, optic atrophy, and recurrent infections. Individuals with partial biotinidase deficiency may have hypotonia, skin rash, and hair loss, particularly during times of stress. All symptomatic children improve when treated with 5 to 10 mg of oral biotin per day.
Natural history without treatment	Prolonged symptoms prior to institution of biotin therapy may leave the patient with varying degrees of neurological sequelae, including mental retardation, seizures, and coma. Death may result from untreated profound biotinidase deficiency.
Natural history with treatment	If treated promptly, biotinidase deficiency may be asymptomatic.
Treatment	Biotin supplement daily
Inheritance	Autosomal recessive
General population incidence	1:60,000 estimated with either profound or partial deficiency
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=253260
Genetests Link	www.geneclinics.org
Support Group	Biotinidase Family Support Group http://biotinidasedeficiency.20m.com/ Children Living with Inherited Metabolic Diseases http://www.climb.org.uk/